Sleep in Individuals with Down Syndrome

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1.
2.
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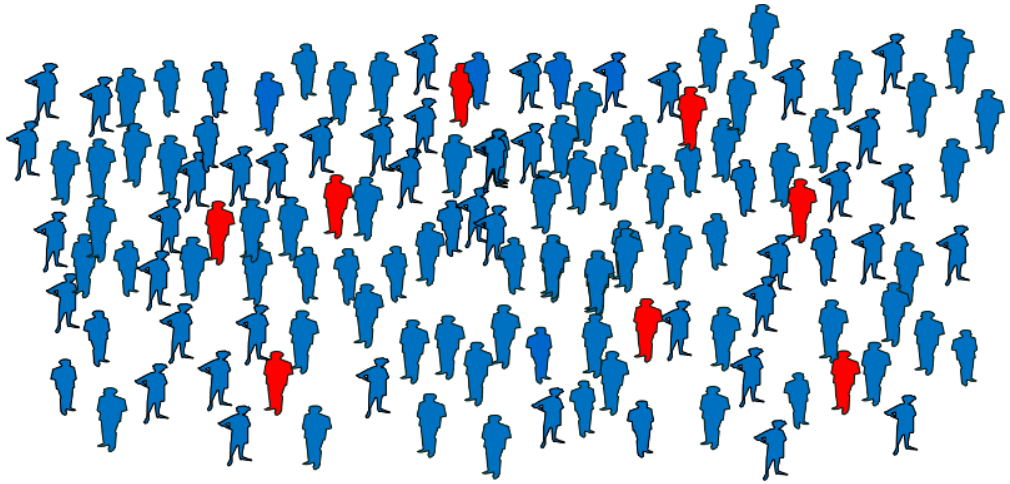
LEARNING OBJECTIVES

At the conclusion of this presentation, attendees should be able to…

1. Identify sleep disorders in individuals with Down syndrome
2. Recognize challenges
3. Provide highlights of ongoing research

OSAS

- OSAS prevalence 45-55%



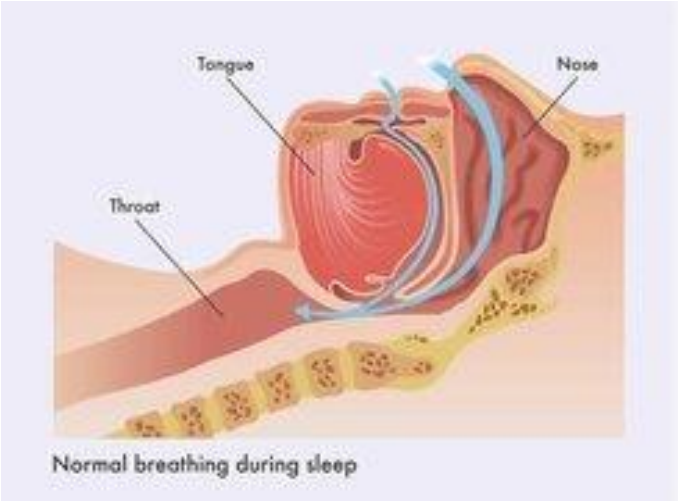
- AAP recommends
  - “evaluation” at age 6 months
  - PSG by age 4 years

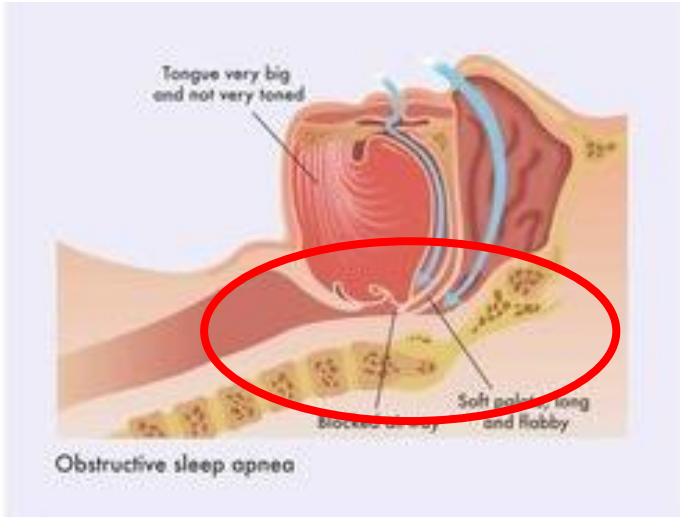
- Many children do not resolve after AT

Pediatrics. 2011 Dec;128(6):1212

Increased OSAS risk

- Midface hypoplasia
- Glossoptosis
- Hypotonia
- Comorbid obesity
- Hypothyroidism



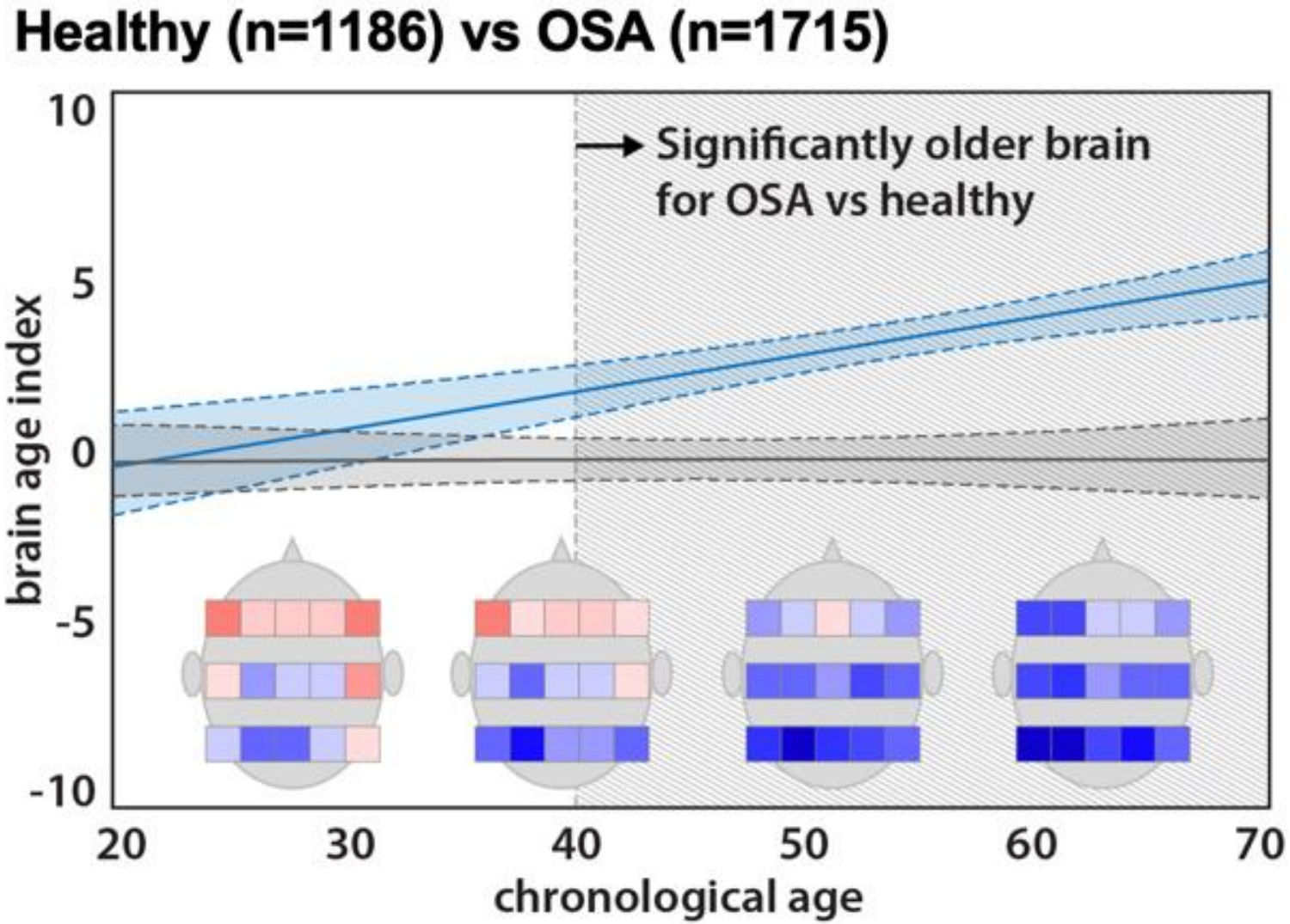


Pediatrics. 1991 Jul;88(1):132-9

Untreated OSAS

- Neurobehavioral deficits
- Systemic hypertension
- Possible elevated cardiometabolic risk
- Possible link with early dementia

OSA and aging



bioRxiv 2022.01.24.477464 https://sites.google.com/usc.edu/nidll

Down syndrome and OSAS

- PSG improves in only a portion of children with DS after AT
- PSG may not be the best outcome
  - Daytime functioning?
  - Quality of life?
  - Cardiometabolic outcomes?
  - Family-centered outcomes?
- Some individuals are referred for PAP initiation

Otolaryngol Head Neck Surg. 2022 Mar;166(3):557-564

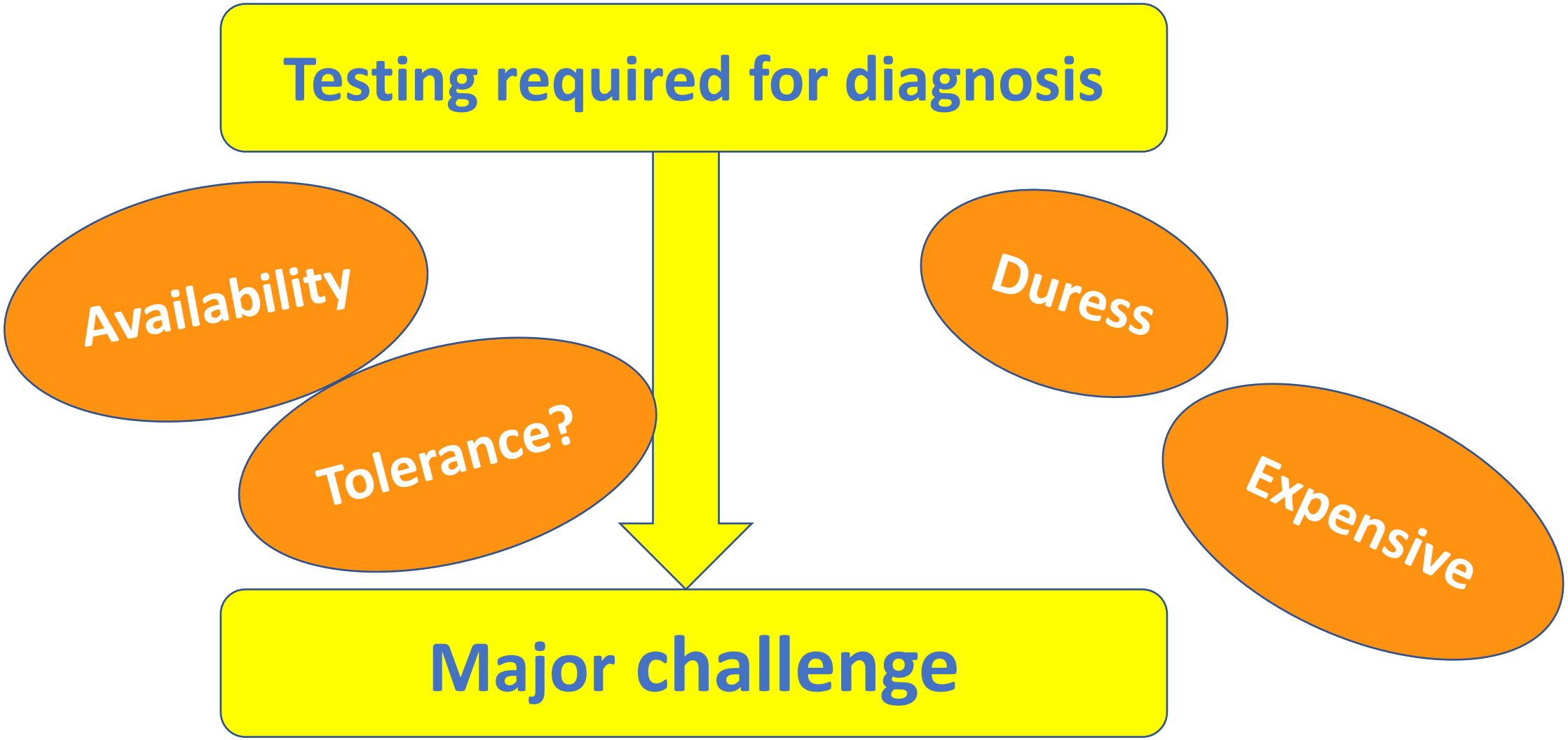
Down syndrome and OSA: The Unknown

- Diagnosis and Treatment of SDB
  - Paucity on in-lab PSG
  - Best screening tool?
  - Role for in-home screening/diagnosis
  - Should adenotonsillectomy be the first line treatment?
  - Lack of RCT

Child Dev. 2015;86:1984-1998
Sleep Breath. 2015;19:1065-1071

Down syndrome and sleep: The Unknown

- Patient-centered outcomes
  - Appropriate neurobehavioral testing?
  - Families may experience different challenges than those of typically developing children?
  - Families may be interested in smaller strides?


Testing required for diagnosis
Availability
Tolerance?
Duress
Expensive
Major challenge

90% of children treated with adenotonsillectomy for presumed OSA never have the diagnosis made by PSG

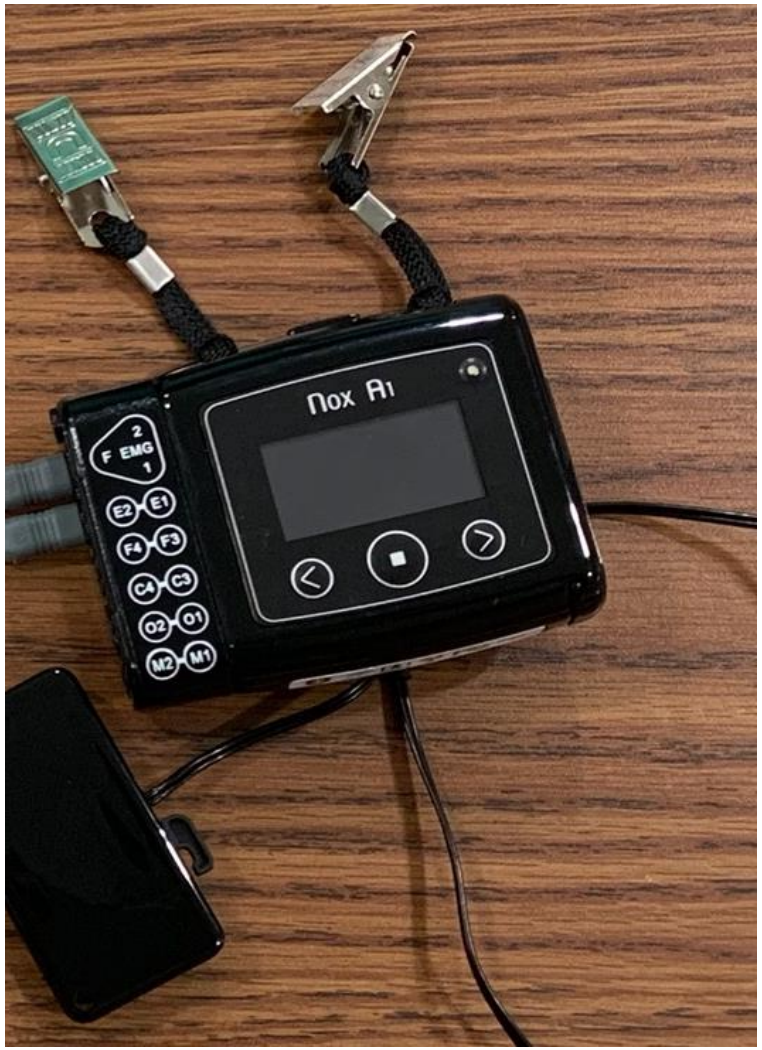
→ Did they really have OSA?

→ Was the surgery needed?

→ Risk/benefit?

Laryngoscope. 2006;116(6):956-958.

R21 HSAT DS



Tolerability, family-reported perceptions/experience

Feasibility

Diagnostic accuracy for moderate-severe OSA

INCLUDE R21 HD101003

N=35

Youth aged 10-20 years

Both in-lab PSG and HSAT in any order

Questionnaires

Videos

- Set up in the hospital: https://www.youtube.com/watch?v=NpDz_I6nxn4

- Set up at home:

https://www.youtube.com/watch?v=vU7st6lBbno

34 tested

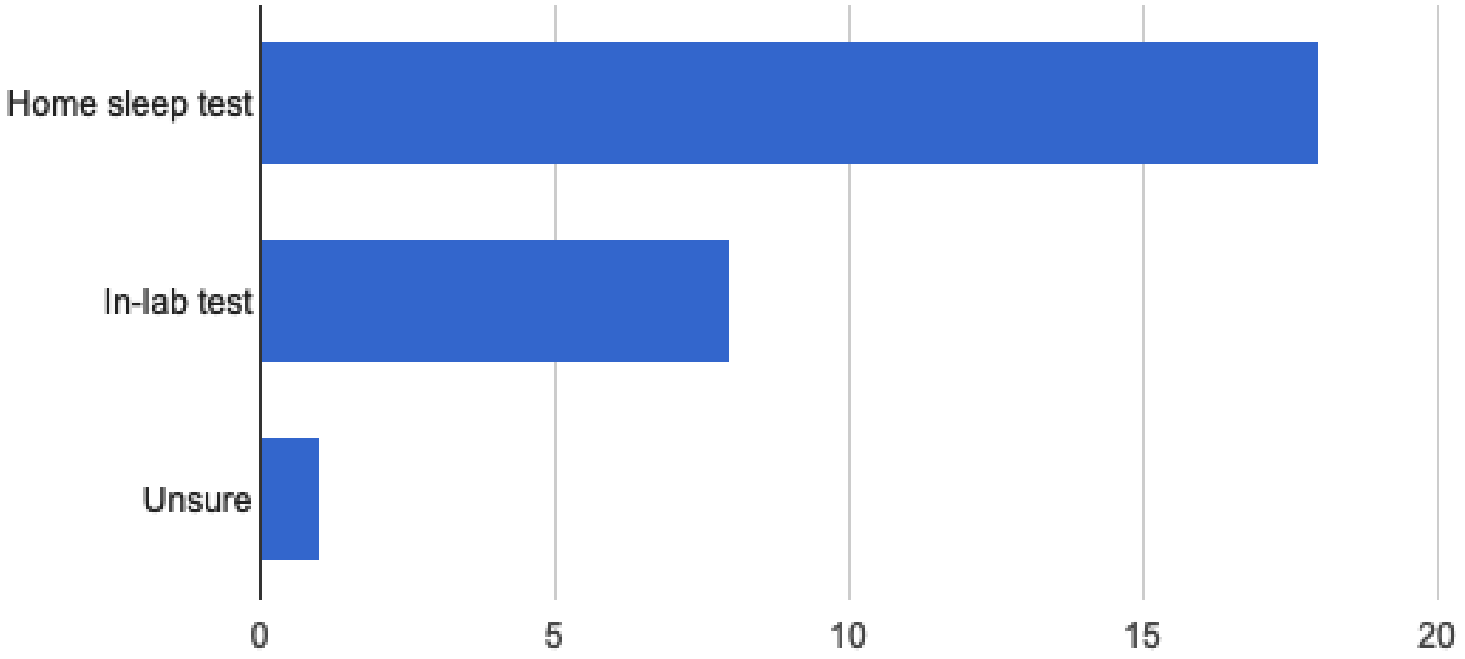
16 [12-18] years

OAHI= 14.2 [4.8-22.8] events per hour

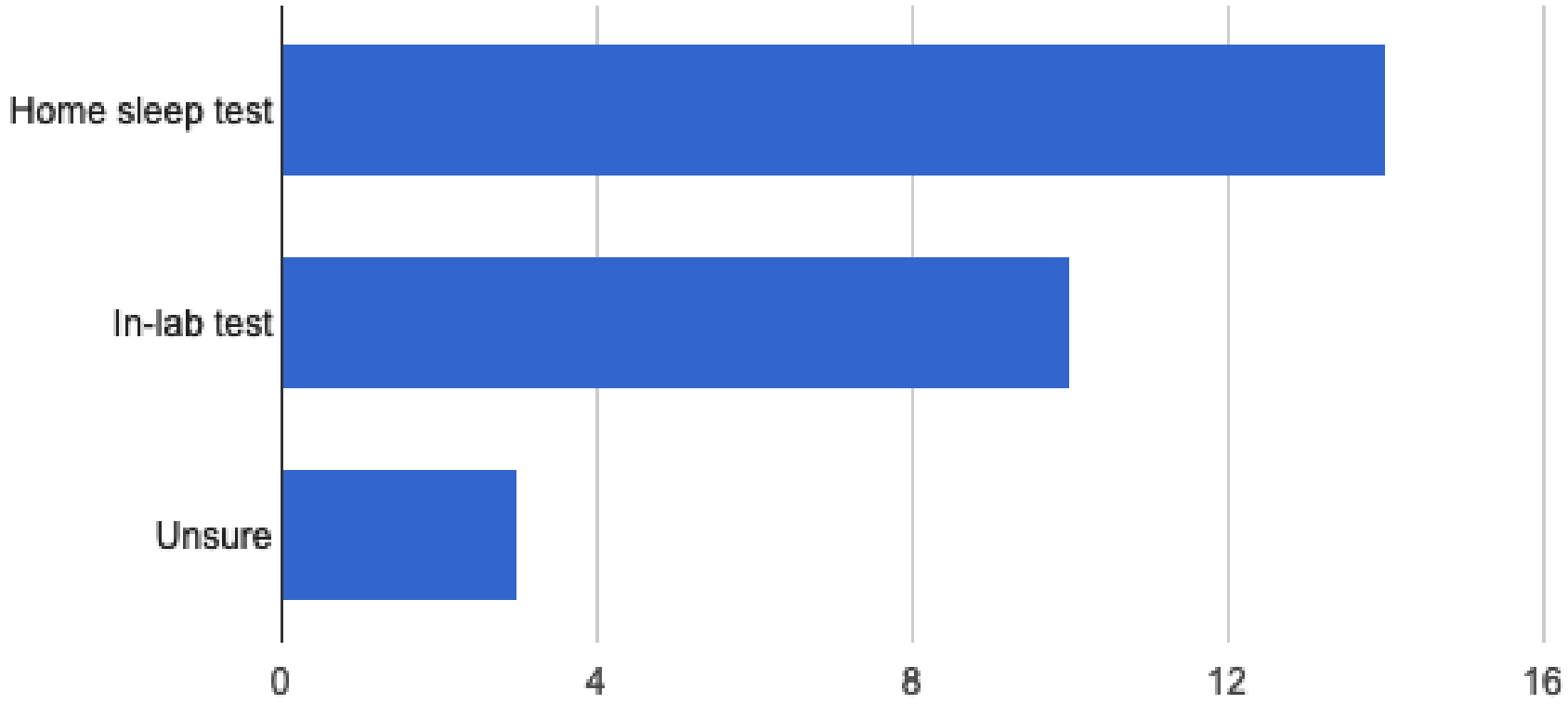
TST = 6.3 [5-7] hours

- < 15 min to set up at home: 94%
- Very easy to place hand oximeter: 100%
- Very easy to set up nasal cannula: 97%
- Anything come off overnight: 63%
- Able to replace it: 94%
- Test seems usable: 97%
- Test seems easy to use: 93%
- Experience was good/very good: 87%

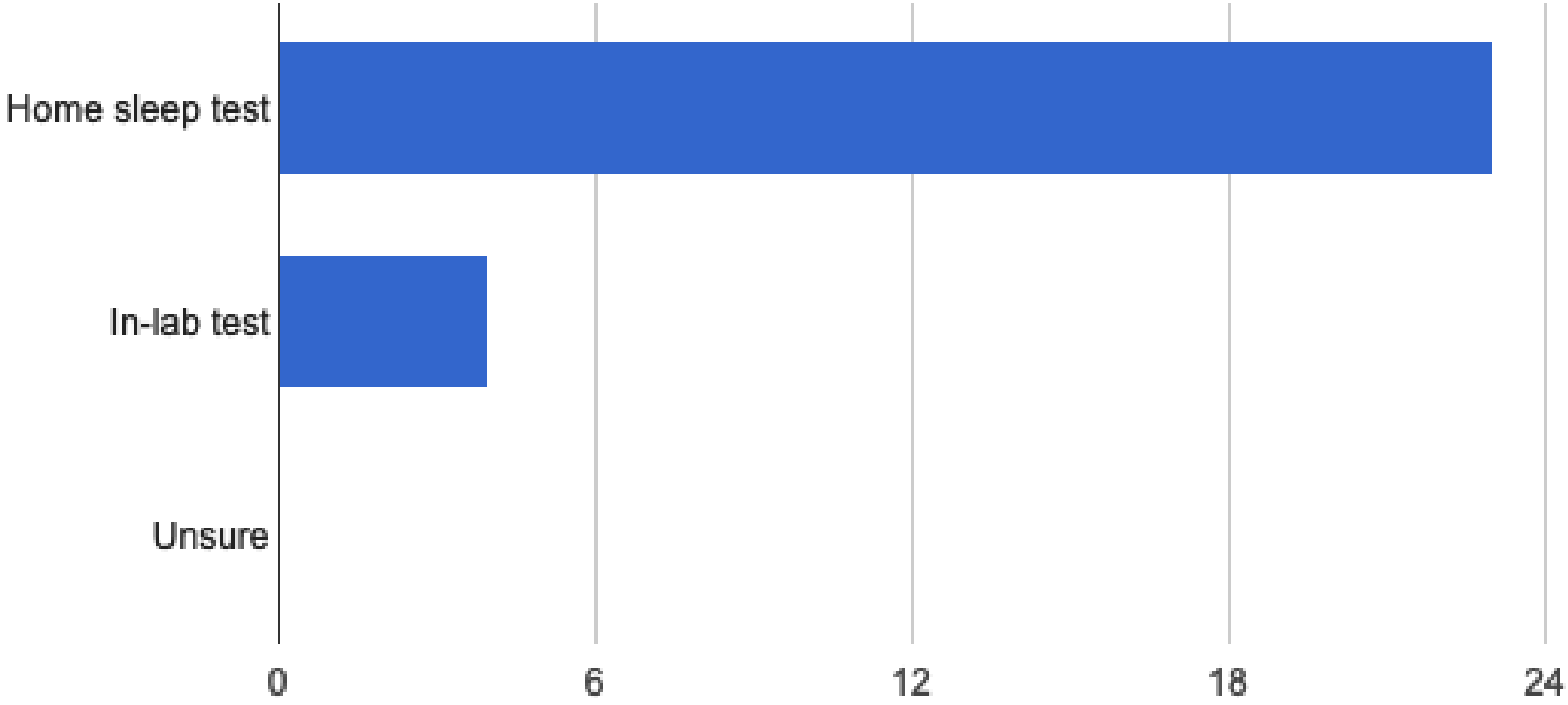
Which test was easier for you?



Which test do you prefer?

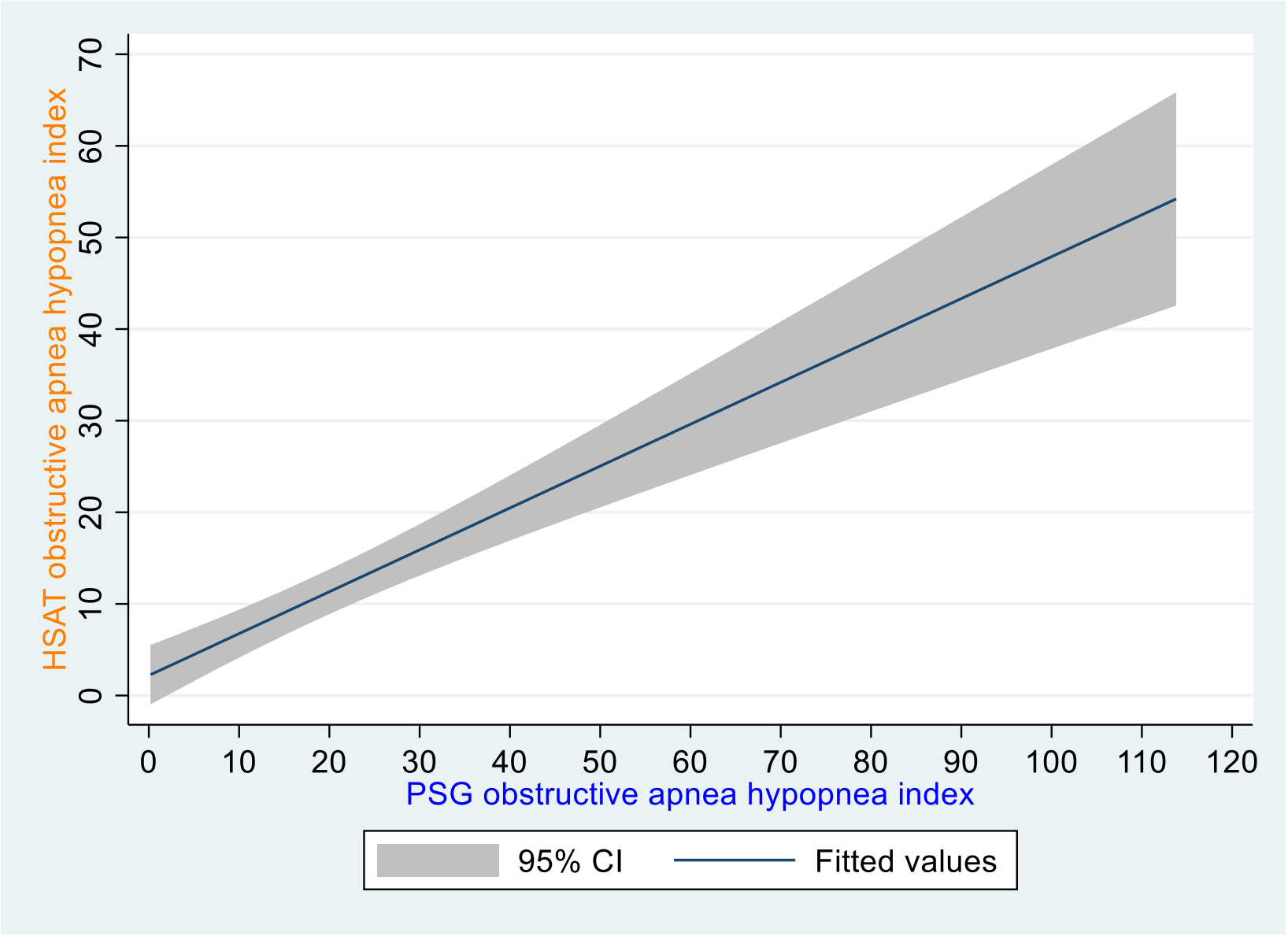


Which test was easier for your child?



HSAT appears to be feasible and well-accepted

Correlation PSG vs HSAT



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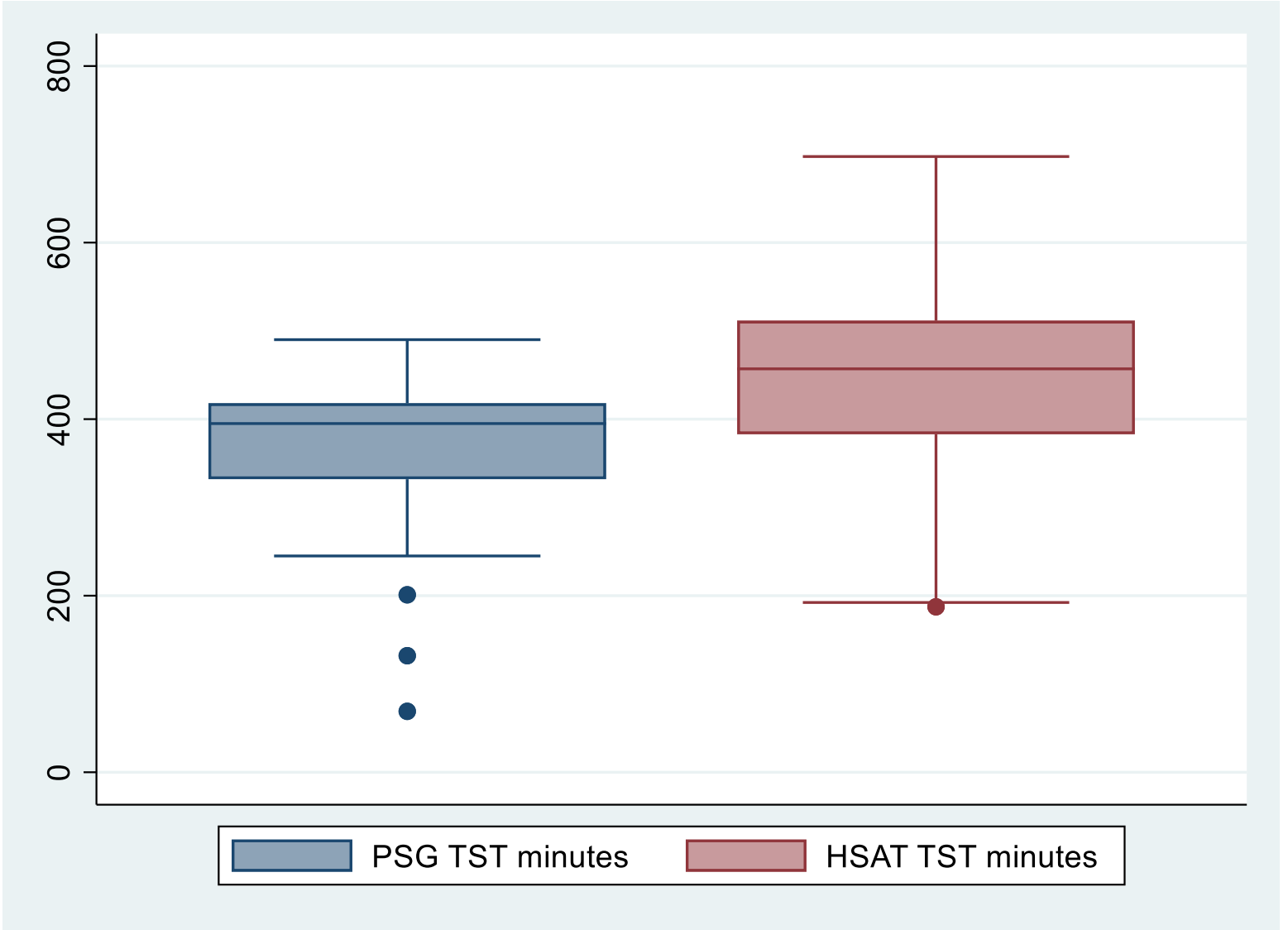
Sensitivity 76.9%

Specificity 88.9%

Total sleep time PSG vs HSAT



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PAP in children with DS

INCLUDE R61/33 HL151253

Strategies to increase PAP use

- Meet families where they are
- Bedtime routine
- Activities that the child enjoys
- Role modeling

Sessions twice per day

- Each session will start with these steps:
  - Begin an activity that is calming and enjoyable for your child. Anything distracting and enjoyable for your child.
  - While the child is complying with requests, provide verbal praise, clapping, cheers, and any other positive reinforcement. Only provide this feedback when your child is doing what you request.
  - Remind the child that only the caregiver (mom, dad, nurse, grandma) takes the mask off.

What if my child has difficulty with the mask/device?

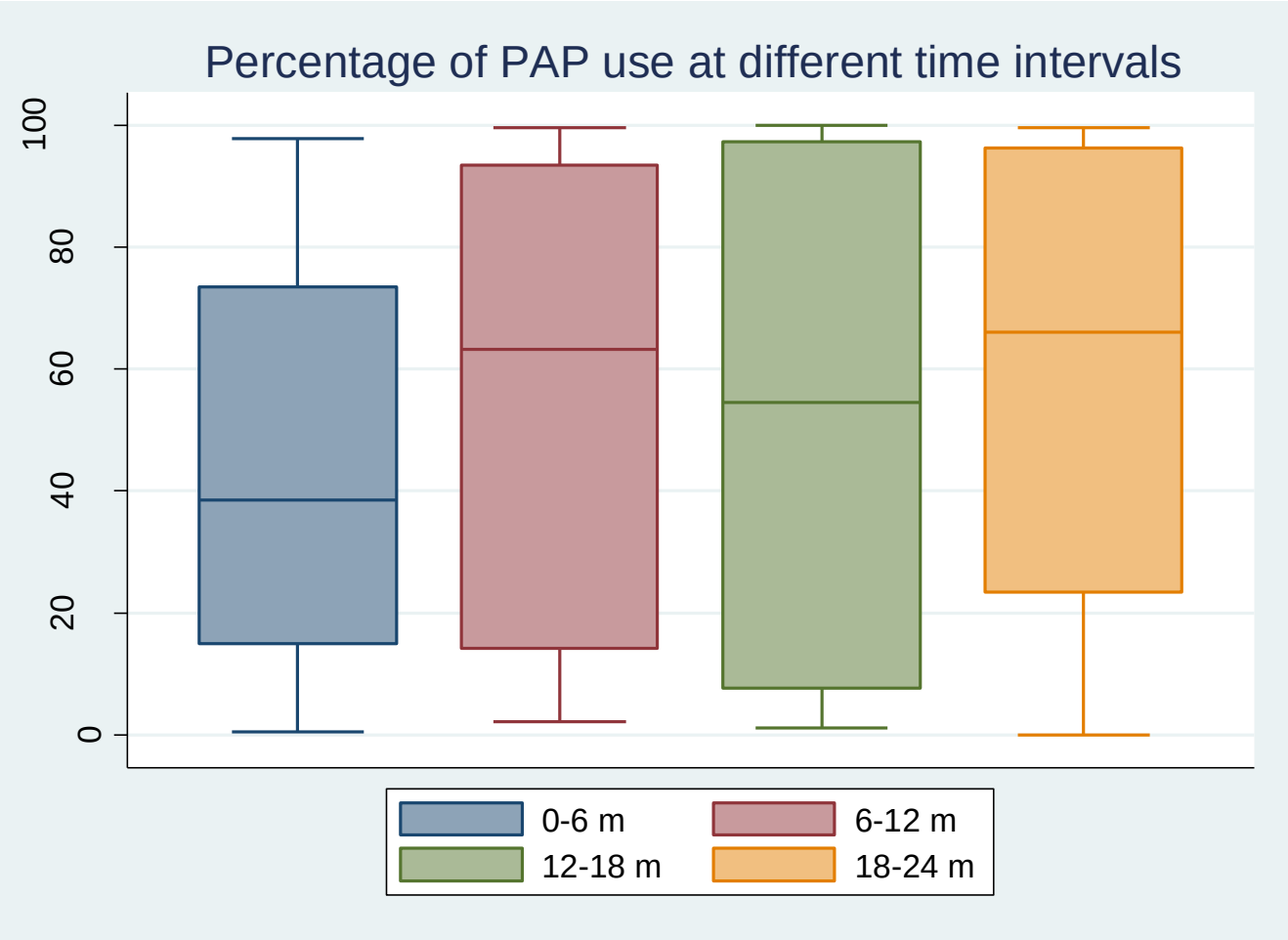
1. Ignore all crying, yelling. Stay calm and positive.
2. Gently guide his/ her hands away from the mask and say “hands down”
3. Repeat the same task until the child is cooperative

Parents qualitative interviews

	High adherence	Low Adherence
Patience	"Just **slow down**, introduce it, **let the child help dictate the timeline**. This is something that's going to be with them forever, so taking those couple of extra days to figure it out. Things to help make it their own and **give them some ownership** of it. Like I said [Child] was **thrilled when we bought ribbo**n, she got to pick out different ribbons. To be able to tie around the tubing. So it's like we might be defacing the machine, but it feels a little bit more like hers then."	"I think you just have to be **patient** with them, patience is definitely I think is the key … my advice to most parents again, you just have patience with them **just figure out a way that will make it fun**, try to say, 'Hey, this is fun, this will help you breathe better,' just interact with them to where it makes it fine."
Visual Supports	"I think children born with **Down syndrome are more visual**. The video, now that I think of it, we did that early. Doing the video and kind of being hands-on with it I think is probably more helpful. That would be just to make sure that you have individuals there for 'em I think helps along with talking to them about it. [Laughs] **Letting them discover some things for themselves** to get comfortable with it instead of pushing something on them."	"I have been kind of thinking of **maybe a little book to write about children with sleep apnea and Down Syndrome** or a little skit or something 'cause if there is one out there, it's hard to find. I felt as though that **they need information on their level that they can understand** because I am still trying to get my daughter to understand … Like I guess as far as what happens to them when they are sleeping. And preventative measures that they can take for those things not to happen**. I guess somebody that looks like them and that is going through the same thing as them**. Someone that they can identify with, I guess."

	High adherence	Low Adherence
Positive Reinforcement	"I think that maybe if there could be some system where you could **encourage parents to have lots of positive reinforcement and fun** like play game or something where 'Let's see how long you can wear it, if you can beat your score from last night,' or something like that. That really helped us anyway 'cause [Child] was then more excited to wear it. Thought more positively instead of almost like a punishment."	"I would definitely say make the mask. Again, **make it fun**. Make it something that they're not scared of … **We got the mask a couple weeks before we ever got the sleep study. We let him play with it, we let him put it on, we let him be an elephant.** Let him just play with it on a day-to-day thing while you're waiting for your BiPAP or CPAP to come in. Let them really just make it part of their day."
Social Support	"Personally, the **whole family has to work at it**. If my husband doesn't help me with it, and it's just me by myself then I would feel overwhelmed, depressed. I would have felt like — at the beginning it was just me or my husband stepped in. **It was still a hassle, but if it was just on me by myself, I couldn't do it.**" "The only little negative side of it is, I've noticed [CPAP] intimidates people. [Child] has **missed out** on some sleepovers with her sister and cousin at her grandparent's house, because it concerns them because it is such a big thing that this controls breathing, and they are very intimidated by that. I think that [Child] **missed out on a school trip because it would've been an overnight and they had issues with the CPAP**. Some of these people, they're experiencing some of the same **thoughts** that we did initially with, '**Oh my gosh this controls breathing, this is a major to-do**.'"	"I think it would be nice to have another parent who was in the same spot. I don't know, **having a child with special needs can sometimes be lonely**. Usually, **you feel like you're the only one that's having to do certain things**." "My **husband looks at it like that's something that I'm dealing with**, so basically, I was in the ER with my [family member] all night and so he didn't put it on her. He said, 'Well, that's your thing.' **Like if I'm not here, it's going to be your thing.** But maybe just making sure that the families are all on the same page."

PAP adherence in children with DS at CHOP



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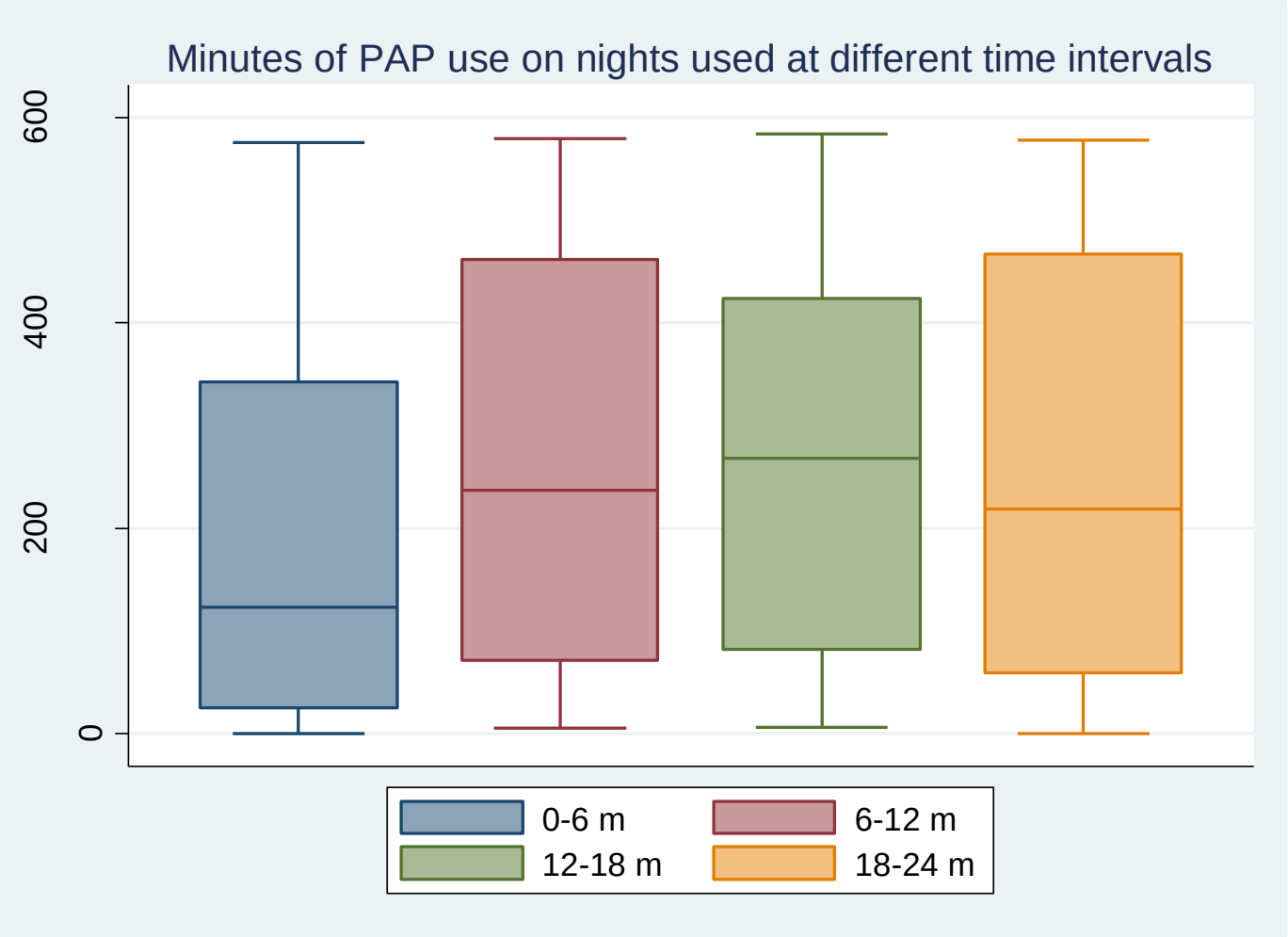
PAP adherence in children with DS at CHOP



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Conclusions

Sleep in individuals with DS has not been well studied

OSAS is very prevalent

Trials of treatment of OSAS are needed

Home Sleep Apnea Testing may have a role in children with DS

Future research

- In-home screening tool: device? Omics? Both?
- RCT of HSAT in individuals with DS
- Data-driven screening algorithm
- Relationship between OSA and aging/Alzheimer in individuals with DS

Thank you!

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Collaborators:

- Andi Kelly, Chris Cielo, Melissa Xanthopoulos
- Michelle Ward, Ahtish Arputhan, Mary Anne Cornaglia, Ruth Bradford
- The Sleep Center @ CHOP